



## SPART gene

spartin

### Normal Function

The *SPART* gene provides instructions for producing a protein called spartin, whose function is not entirely understood. Researchers believe that spartin may be involved in a variety of cell functions, from breaking down proteins to transporting materials from the cell surface into the cell (endocytosis). Spartin is found in a wide range of body tissues, including the nervous system.

### Health Conditions Related to Genetic Changes

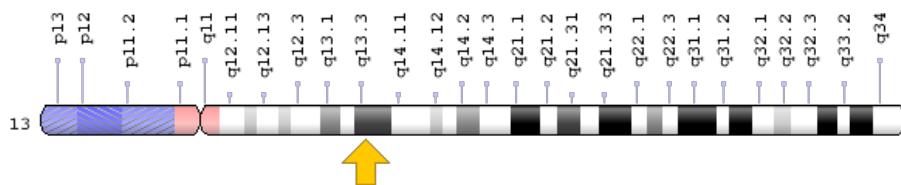
#### Troyer syndrome

One mutation has been identified in people with Troyer syndrome. This mutation deletes a DNA building block (nucleotide) in the *SPART* gene. Specifically, this mutation deletes the nucleotide adenosine at position 1110 in the gene, written as 1110delA. This mutation results in an abnormally shortened, nonfunctioning spartin protein. It is unclear how this mutation causes the signs and symptoms of Troyer syndrome.

### Chromosomal Location

Cytogenetic Location: 13q13.3, which is the long (q) arm of chromosome 13 at position 13.3

Molecular Location: base pairs 36,301,638 to 36,370,180 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## **Other Names for This Gene**

- KIAA0610
- SPARTIN
- spastic paraplegia 20
- spastic paraplegia 20 (Troyer syndrome)
- spastic paraplegia 20, spartin (Troyer syndrome)
- SPG20
- SPG20\_HUMAN
- TAHCCP1
- trans-activated by hepatitis C virus core protein 1

## **Additional Information & Resources**

### GeneReviews

- Troyer Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1382>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SPG20%29+OR+%28SPART%29%29+OR+%28spartin%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- SPG20 GENE  
<http://omim.org/entry/607111>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SPG20.html](http://atlasgeneticsoncology.org/Genes/GC_SPG20.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SPART%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=18514](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18514)

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/23111>
- UniProt  
<http://www.uniprot.org/uniprot/Q8N0X7>

## Sources for This Summary

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